AMENDMENTS TO THE CLAIMS

This listing of claims will replace all prior versions, and listings, of claims in the application.

What is claimed is:

1. (previously presented) An isolated nucleic acid molecule at least 20 nucleotides in length, wherein the nucleic acid shares at least 95% sequence identity with a corresponding sequence from SEQ ID NO:1 or SEQ ID NO:2.

2. (currently amended) The nucleic acid of claim 1, ~~wherein the nucleic acid is a gene comprising a~~ comprising a mutation relative to SEQ ID NO:1 or SEQ ID NO:2.

3. (currently amended) The nucleic acid of claim 2, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 of ~~(~~SEQ ID NO: 1~~)~~; (b) a deletion from nucleotide 511 to nucleotide 6944 of ~~(~~SEQ ID NO: 1~~)~~; (c) an insertion of T between nucleotide numbers 1334 and 1335 of ~~(~~SEQ ID NO: 2~~)~~; (d) a deletion of CTT spanning nucleotides 1346-1348 of ~~(~~SEQ ID NO: 2~~)~~; (e) an A to G substitution ~~a~~ at position 9107 of ~~(~~SEQ ID NO: 1~~)~~; (f) a G to T substitution at position 1461 of ~~(~~SEQ ID NO: 2~~)~~; (g) a C to T substitution at position 429 of ~~(~~SEQ ID NO: 2~~)~~; (h) a G to T substitution at position 1209 of ~~(~~SEQ ID NO: 2~~)~~; (i) a CC deletion at 598-599 of ~~(~~SEQ ID NO: 2~~)~~; and (j) a C to T substitution at position 639 of ~~(~~SEQ ID NO: 2~~)~~.

4. (previously presented) The nucleic acid of claim 2, wherein the mutation is associated with development of mucolipidosis IV.

5. (previously presented) The nucleic acid of claim 1, which encodes a polypeptide having an amino acid sequence at least 95% identical to SEQ ID NO: 3.

6. (original) The nucleic acid of claim 5, wherein the polypeptide has an amino acid sequence as depicted in SEQ ID NO:3.

7. (original) The nucleic acid of claim 6 which has a nucleotide sequence as depicted in SEQ ID NO:1 or SEQ ID NO:2.

Claims 8-11 are cancelled.

12. (currently amended) A method for detecting a mutation associated with a mucolipidosis, which method comprises ~~in a mammal, which method comprises using an oligonucleotide of claim 39 to detect a mutation in a gene having a sequence at least 95% identical to SEQ ID NO: 1.~~

(a) contacting a sample suspected of comprising a nucleic acid having a sequence at least 95% identical to SEQ ID NO: 1 with an oligonucleotide of claim 39 under conditions that permit hybridization of the oligonucleotide to the nucleic acid, and

(b) determining the presence of a hybrid formed between the oligonucleotide and the nucleic acid; wherein said determining indicates the presence or absence of a mutation in the nucleic acid.

13. (currently amended) The method according to claim 12, wherein the mutation consists of an insertion in the nucleic acid ~~gene~~.

14. (currently amended) The method according to claim 12, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 of ~~(~~SEQ ID NO: 1~~)~~; (b) a deletion from nucleotide 511 to nucleotide 6944 of ~~(~~SEQ ID NO: 1~~)~~; (c) an insertion of T between nucleotide numbers 1334 and 1335 of ~~(~~SEQ ID NO: 2~~)~~; (d) a deletion of CTT spanning nucleotides 1346-1348 of ~~(~~SEQ ID NO: 2~~)~~; (e) an A to G substitution ~~a~~ at position 9107 of ~~(~~SEQ ID NO: 1~~)~~; (f) a G to T substitution at position 1461 of ~~(~~SEQ ID NO: 2~~)~~; (g) a C to T substitution at position 429 of ~~(~~SEQ ID NO: 2~~)~~; (h) a G to T substitution at position 1209 of ~~(~~SEQ ID NO: 2~~)~~; (i) a CC deletion at 598-599 of ~~(~~SEQ ID NO: 2~~)~~; and (j) a C to T substitution at position 639 of ~~(~~SEQ ID NO: 2~~)~~.

15. (withdrawn) The method according to claim 12, wherein the mucolipidosis is mucolipidosis IV.

16. (currently amended) A method for diagnosing a mucolipidosis, which method comprises ~~using an oligonucleotide of claim 39 to detect a mutation in a having a sequence at least 95% identical to SEQ ID NO: 1, wherein the mutation is associated with development of mucolipidosis.~~

(a) contacting a sample suspected of comprising a nucleic acid having a sequence at least 95% identical to SEQ ID NO: 1 with an oligonucleotide of claim 39 under conditions that permit hybridization of the oligonucleotide to the nucleic acid, and

(b) determining the presence of a hybrid formed between the oligonucleotide and the nucleic acid; wherein said determining indicates the presence or absence of a mutation in the nucleic acid.

17. (currently amended) The method according to claim 16, wherein the mutation is selected from the group consisting of an insertion in the nucleic acid ~~gene~~, a deletion of the nucleic acid ~~gene~~, a truncation of the nucleic acid ~~gene~~, a nonsense mutation, a frameshift mutation, a splice-site mutation, and a missense mutation.

18. (currently amended) The method according to claim 16, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 of ~~(~~SEQ ID NO: 1~~)~~; (b) a deletion from nucleotide 511 to nucleotide 6944 of ~~(~~SEQ ID NO: 1~~)~~; (c) an insertion of T between nucleotide numbers 1334 and 1335 of ~~(~~SEQ ID NO: 2~~)~~; (d) a deletion of CTT spanning nucleotides 1346-1348 of ~~(~~SEQ ID NO: 2~~)~~; (e) an A to G substitution ~~a~~ at position 9107 of ~~(~~SEQ ID NO: 1~~)~~; (f) a G to T substitution at position 1461 of ~~(~~SEQ ID NO: 2~~)~~; (g) a C to T substitution at position 429 of ~~(~~SEQ ID NO: 2~~)~~; (h) a G to T substitution at position 1209 of ~~(~~SEQ ID NO: 2~~)~~; (i) a CC deletion at 598-599 of ~~(~~SEQ ID NO: 2~~)~~; and (j) a C to T substitution at position 639 of ~~(~~SEQ ID NO: 2~~)~~.

19. (original) The method according to claim 16, wherein the mucolipidosis is MLIV.

20. (currently amended) A method for predicting the likelihood of developing MLIV, which method comprises ~~comprising using an oligonucleotide of claim 39 to detect a mutation in a gene associated~~

~~with development of mucolipidosis IV, having at least 95% sequence identity to SEQ ID NO:1 and determining that there is a likelihood of developing MLIV if the mutation is present.~~

(a) contacting a sample suspected of comprising a nucleic acid having a sequence at least 95% identical to SEQ ID NO: 1 with an oligonucleotide of claim 39 under conditions that permit hybridization of the oligonucleotide to the nucleic acid;

(b) determining the presence of a hybrid formed between the oligonucleotide and the nucleic acid, wherein said determining indicates the presence or absence of a mutation in the nucleic acid; and

(c) establishing that there is a likelihood of developing MLIV if the mutation is present.

21. (previously presented) The method according to claim 20, wherein the mutation consists of an insertion in the gene.

22. (currently amended) The method according to claim 20, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 of ~~(~~SEQ ID NO: 1~~)~~; (b) a deletion from nucleotide 511 to nucleotide 6944 of ~~(~~SEQ ID NO: 1~~)~~; (c) an insertion of T between nucleotide numbers 1334 and 1335 of ~~(~~SEQ ID NO: 2~~)~~; (d) a deletion of CTT spanning nucleotides 1346-1348 of ~~(~~SEQ ID NO: 2~~)~~; (e) an A to G substitution ~~a~~ at position 9107 of ~~(~~SEQ ID NO: 1~~)~~; (f) a G to T substitution at position 1461 of ~~(~~SEQ ID NO: 2~~)~~; (g) a C to T substitution at position 429 of ~~(~~SEQ ID NO: 2~~)~~; (h) a G to T substitution at position 1209 of ~~(~~SEQ ID NO: 2~~)~~; (i) a CC deletion at 598-599 of ~~(~~SEQ ID NO: 2~~)~~; and (j) a C to T substitution at position 639 of ~~(~~SEQ ID NO: 2~~)~~.

23. (currently amended) A kit for detecting a mutation in a ~~gene~~ nucleic acid having at least 95% sequence identity to SEQ ID NO:1, comprising an oligonucleotide of claim 39 that specifically hybridizes to or adjacent to the mutation, and a means for detecting binding of the oligonucleotide to the nucleic acid.

24. (previously presented) The kit according to claim 23, wherein the oligonucleotide is a labeled probe.

25. (previously presented) The kit according to claim 23, wherein the oligonucleotide hybridizes to a first site adjacent to the mutation, further comprising a second oligonucleotide that specifically hybridizes to a second site adjacent to the mutation, wherein the second site is on the opposite strand relative to the first site, and oriented relative to the first site such that both sites flank opposite sides of the site of the mutation, whereby the first and second oligonucleotides serve as primers for PCR amplification of the site of the mutation.

26. (currently amended) The kit according to claim 23, wherein the mutation is selected from the group consisting of an insertion in the nucleic acid ~~gene~~, a deletion of the nucleic acid ~~gene~~, a truncation of the nucleic acid ~~gene~~, a nonsense mutation, a frameshift mutation, a splice-site mutation, and a missense mutation.

27. (currently amended) The kit according to claim ~~26~~ 23, wherein the mutation is selected from the group consisting of: (a) an A to G substitution at position 5534 of ~~(~~SEQ ID NO: 1~~)~~; (b) a deletion

from nucleotide 511 to nucleotide 6944 of ~~(~~SEQ ID NO: 1~~)~~; (c) an insertion of T between nucleotide numbers 1334 and 1335 of ~~(~~SEQ ID NO: 2~~)~~; (d) a deletion of CTT spanning nucleotides 1346-1348 of ~~(~~SEQ ID NO: 2~~)~~; (e) an A to G substitution ~~a~~ at position 9107 of ~~(~~SEQ ID NO: 1~~)~~; (f) a G to T substitution at position 1461 of ~~(~~SEQ ID NO: 2~~)~~; (g) a C to T substitution at position 429 of ~~(~~SEQ ID NO: 2~~)~~; (h) a G to T substitution at position 1209 of ~~(~~SEQ ID NO: 2~~)~~; (i) a CC deletion at 598-599 of ~~(~~SEQ ID NO: 2~~)~~; and (j) a C to T substitution at position 639 of ~~(~~SEQ ID NO: 2~~)~~.

Claims 28-32 are cancelled.

33. (previously presented) An expression vector comprising the nucleic acid of claim 5, operatively associated with a promoter.

34. (previously presented) The expression vector of claim 33, wherein the nucleic acid encodes the amino acid sequence as depicted in SEQ ID NO:3.

35. (currently amended) A ~~pharmaceutical composition comprising the expression~~ vector ~~of~~ comprising the nucleic acid of claim 1. ~~33 and a pharmaceutically acceptable carrier or excipient.~~

Claims 36-38 are cancelled.

39. (previously presented) The nucleic acid of claim 1, wherein the nucleic acid is a single stranded oligonucleotide.

40. (currently amended) The nucleic acid of claim 2, wherein the mutation is an insertion in the ~~gene~~ nucleic acid.

41. (currently amended) The nucleic acid of claim 2, wherein the mutation is a deletion of the ~~gene~~ nucleic acid.

42. (previously presented) The nucleic acid of claim 2, wherein the mutation is a point-mutation.

43. (previously presented) The nucleic acid of claim 2, wherein the mutation is a nonsense mutation.

44. (previously presented) The nucleic acid of claim 2, wherein the mutation is a frameshift mutation.

45. (previously presented) The nucleic acid of claim 2, wherein the mutation is a missense mutation.

46. (previously presented) The nucleic acid of claim 2, wherein the mutation is an mRNA splicing mutation.

47. (currently amended) The method according to claim 12, wherein the mutation is a deletion of the ~~gene~~ nucleic acid.

48. (previously presented) The method according to claim 12, wherein the mutation is a point-mutation.

49. (previously presented) The method according to claim 12, wherein the mutation is a nonsense mutation.

50. (previously presented) The method according to claim 12, wherein the mutation is a frameshift mutation.

51. (previously presented) The method according to claim 12, wherein the mutation is a missense mutation.

52. (previously presented) The method according to claim 12, wherein the mutation is an mRNA splicing mutation.

53. (currently amended) The method according to claim 20, wherein the mutation is a deletion of the ~~gene~~ nucleic acid.

54. (previously presented) The method according to claim 20, wherein the mutation is a point-mutation.

55. (previously presented) The method according to claim 20, wherein the mutation is a nonsense mutation.

56. (previously presented) The method according to claim 20, wherein the mutation is a frameshift mutation.

57. (previously presented) The method according to claim 20, wherein the mutation is a missense mutation.

58. (previously presented) The method according to claim 20, wherein the mutation is an mRNA splicing mutation.